Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/9/1988
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Amniotic Fluid

ARUP test code 2002293

Chromosome Analysis, Amniotic Fluid

See Note (Ref Interval: Normal)

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Test performed:      Chromosome Analysis
Specimen type:       Amniotic Fluid
Reason for referral: Abnormal maternal serum screen for Trisomy
21, Abnormal NIPT for Trisomy 21

Laboratory analysis

Number of cells counted:    21
Number of colonies counted: 8
Number of cells analyzed:   21
Number of cells karyotyped: 21
ISCN Band level:            400
Banding Method:             G-Banding
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RESULT
Abnormal Karyotype (Male)

Trisomy 21 (Down syndrome)

47,XY,+21
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INTERPRETATION
This analysis showed an additional copy (trisomy) of chromosome
21 in each metaphase.

This result is consistent with a clinical diagnosis of trisomy
21 (Down syndrome). Features associated with Down syndrome may
include hypotonia, a characteristic facial appearance,
developmental delays / intellectual disability, and short
stature. Other findings may include congenital abnormalities of
the heart, diaphragm, and digestive tract, hypothyroidism, and
hearing and vision difficulties. Minor physical anomalies
commonly seen in individuals with Down syndrome include single
transverse palmar crease, low-set ears, and sandal-gap toe. Not
all fetuses with Down syndrome will have ultrasound anomalies.
Life span may be shortened as compared to the general
population, and there is an increased risk of developing
leukemia and Alzheimer's disease later in life.

NOTE: FISH was performed on this sample and reported under ARUP
accession #(23-037-119175). FISH results were ABNORMAL.

No other abnormalities were detected. The standard cytogenetic
methodology used in this analysis may not detect small
rearrangements or low-level mosaicism and cannot detect
submicroscopic deletions or duplications that are detectable by
genomic microarray analysis.

Recommendation:
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H=High, L=Low, *=Abnormal, C=Critical

Genetic counseling

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

References:
1) Sheets et al. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. J Genet Couns. 2011 Oct;20(5):432-41. PMID: 21618060.
2) Bull. Committee on Genetics. Health supervision for children with Down syndrome. Pediatrics. 2011 Aug;128(2):393-406. PMID: 21788214.
3) Jones et al. Smith's Recognizable Patterns of Human Malformations. 7th edition. Philadelphia, PA: Elsevier Saunders; 2013:7-13.

This result has been reviewed and approved by [redacted]

A portion of this analysis was performed at the following location(s):
[redacted]

INTERPRETIVE INFORMATION: Chromosome Analysis, Amniotic Fluid

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

EER Chromosome Analysis Amniotic Fluid	See Note Authorized individuals can access the ARUP Enhanced Report using the following link: [redacted]

Chromosome FISH, Amniotic Fluid with Reflex to Chromosome Analysis or Genomic Microarray

ARUP test code 2011130

Chromosome FISH, Prenatal	See Note	(Ref Interval: Normal)

H=High, L=Low, *=Abnormal, C=Critical

Test Performed: Amniotic Fluid FISH Reflex (AF F RFLX)
Specimen Type: Direct (uncultured) amniocytes
Indication for Testing: Abnormal maternal serum and Abnormal NIPT for Trisomy 21

RESULT
Abnormal FISH Result (Male)

Trisomy 21

This specimen is being reflexed to chromosome analysis.

INTERPRETATION
This analysis showed three hybridization signals for chromosome 21, consistent with trisomy 21.

Aneuploidy of other chromosomes, structural abnormalities, and mosaicism have not been ruled out by this analysis. According to ACMG guidelines, clinical decision-making should not be based on the result of this test alone. Additional testing is recommended for the final interpretation of this result; pending results will be reported separately.

NOTE: Interphase FISH analysis cannot provide structural information accounting for this gain. Please refer to the pending chromosome analysis for further characterization of the structure of this finding.

This analysis was performed with chromosome enumeration probes for 13, 18, 21, X and Y using the FDA-approved AneuVysion probe kit (Abbott Molecular). A total of 50 interphase cells were scored for each probe.

Recommendation:
Genetic counseling

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

Cytogenomic Nomenclature (ISCN):
nuc ish(DXZ1x1,DYZ3x1,D18Z1x2),(RB1x2,D21S259/D21S341/D21S342x3)

This result has been reviewed and approved by [redacted]

A portion of this analysis was performed at the following location(s):
[redacted]

INTERPRETIVE INFORMATION: Chromosome Analysis,
Prenatal FISH

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Amniotic Fluid	23-037-119175	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome Analysis Amniotic Fluid	23-037-119175	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Chromosome FISH, Prenatal	23-037-119175	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical